

MGI (Jackson Laboratory) Mouse Anatomy Use Case examples
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Use Case 1	Annotating to Adult Mouse Anatomy Vocabulary – 30K feet
Case:	Annotate the location of an expression result in an adult mouse
Primary Actor:	Expression curator
Background:	The curator needs to record an gene expression result for an anatomical structure
Problem:	<p>The curator</p> <ol style="list-style-type: none"> 1) selects anatomical structure S from a browser (or types part S to get a list of possible structures and selects one) <p>The system records the result.</p> <p><i>Question: how much of this should focus on the requirements of the system vs. the requirements of the vocabulary? As stated this doesn't tell us much about the vocabulary.</i></p>
Solution:	

Use Case 2	Annotating to Adult Mouse Anatomy Vocabulary – more detail
Case:	Annotate the location of an expression result in an adult mouse
Primary Actor:	Expression curator
Background:	<p>The curator has a result in hand of the form:</p> <p>Assay type A using probe P detects expression of gene G in anatomical structure S in genotype (strain) T</p> <p>and needs to record this result.</p>
Problem:	<p>The curator</p> <ol style="list-style-type: none"> 2) enters assay type A (ignore details) assay types are: Immunohistochemistry, RNA in situ, RNA in situ reporter (knock-in), Northern blot, Western blot, RNase protection, Nuclease S1, RT-PCR 3) probe P (ignore details) 4) enters gene G (ignore details) 5) enters genotype (strain) T (ignore details) 6) selects anatomical structure S from a browser (or types part S to get a list of possible structures and selects one) <p>The system records the result.</p>
Solution:	<ol style="list-style-type: none"> 1) the anatomy must support different levels of spatial resolution to support the different assay types. Immunohistochemistry and RNA in situ can be very high resolution, Northern and Western blot are low resolution. (the point is to describe expression data at different levels of resolution and integrate them all) 2) still doesn't tell us much about the vocabulary

Use Case 3	Query using Adult Mouse Anatomy Vocabulary – 30K feet
Case:	Query for expression results in an adult mouse
Primary Actor:	Researcher
Background:	Researcher wants to know genes are expressed in a specific anatomical structure (or substructure).
Problem:	<p>Researcher selects a structure from an anatomy browser OR types some part of an anatomy term to get a list of possible structures and selects one.</p> <p>System returns all expression results annotated to that structure or any substructures.</p> <p><i>Question: talk about other possible query parameters? Again, how much do we focus on the system vs. the vocabulary?</i></p>
Solution:	<ol style="list-style-type: none"> 1) the anatomy must be hierarchical so that queries for structures can return results for substructures 2) synonyms must be supported since the user can type anatomy terms

Use Case 4	Annotating to Adult Mouse Anatomy Vocabulary – Sampling issue
Case:	Annotate the location of an expression result in an adult mouse – where the structure is not very specific
Primary Actor:	Expression curator
Background:	<p>The curator has a result in hand of the form:</p> <p>Assay type A using probe P detects expression of gene G in anatomical structure S in genotype (strain) T</p> <p>But structure S is not very specific. Examples:</p> <ul style="list-style-type: none"> • just “lung” instead of “left lung” or “right lung” • just “alveolus” instead of “left lung alveolus” or “right lung alveolus” <p>and needs to record this result.</p>
Problem:	<p>The curator (same above?)</p> <ol style="list-style-type: none"> 7) enters assay type A (ignore details) assay types are: Immunohistochemistry, RNA in situ, RNA in situ reporter (knock-in), Northern blot, Western blot, RNase protection, Nuclease S1, RT-PCR 8) probe P (ignore details) 9) enters gene G (ignore details) 10) enters genotype (strain) T (ignore details) 11) selects anatomical structure S from a browser (or types part S to get a list of possible structures and selects one) <p>The system records the result.</p>
Solution:	<ol style="list-style-type: none"> 1) the anatomy must support different levels of genericity. Curators need to be able to add more generic structures as needed.

Use Case 5	Querying using Adult Mouse Anatomy Vocabulary – Multiple relationships
Case:	Query for expression results in an adult mouse – but the same structure can be viewed as a substructure of different parents
Primary Actor:	Researcher
Background:	Researcher wants to know genes are expressed in a specific anatomical structure (or substructure).
Problem:	<p>Researcher selects a structure from an anatomy browser, say “sensory organ system” Results annotated to “eye” should be returned.</p> <p>OR</p> <p>Researcher selects a structure from an anatomy browser, say “head” Again, results annotated to “eye” should be returned.</p>
Solution:	1) the vocabulary must support a DAG structure.

Other Issues not addressed by the above Use Cases:

- 1) entering negative expression results
- 2) querying for negative expression results
- 3) updating the anatomy, say to add more detailed substructures or add more generic structures, adding synonyms, etc.
- 4) browsing the anatomy
- 5) where to define vocabulary boundaries, e.g., does the anatomy include cell types?
- 6) what to do about describing abnormal anatomy, e.g., extra digits
- 7) the anatomy should be used in phenotype descriptions (e.g., abnormal development of X), including cancer phenotypes

Our one concrete thought about all this:

- 1) Use cases that are intended to guide vocabulary development (or selection of existing vocabularies) should include as many actual examples as is feasible, particularly around obviously sticky areas.